

CLAIMS:

5 *sub C8* 1. A method for detecting the presence or absence of twelve mutations in the cystic fibrosis transmembrane conductor regulator (CFTR) gene, which method comprises contacting sample genomic DNA from an individual in two separate reaction vessels with allele specific primer sets for (A) 1717-1 G>A, G542X, W1282X, N1303K, ΔF508(M), 3849+10kb C>T mutations and (B) the 621+1 G>T, R553X, G551D, R117H, R1162X and 10 R334W mutations respectively, in the presence of appropriate nucleotide triphosphates and an agent for polymerisation, such that each diagnostic primer is extended only when the relevant mutation is present in the sample; and detecting the presence or absence of CFTR gene alleles by reference to the presence or absence of diagnostic primer extension product(s).

15 *sub C2* 2. A method as claimed in claim 1 and wherein one or more diagnostic primers is used with one or more amplification primers in one or more cycles of PCR amplification.

20 *sub C2* 3. A set of allele specific primers for each of the following alleles of the CFTR gene: 1717-1 G>A, G542X, W1282X, N1303K, ΔF508(M), and 3849+10kb C>T *mutations*.

4. A set of primers as claimed in claim 3 and comprising the following diagnostic primer sequences:

TCTGGGATTCAATAACGTTGCAACAGTCA
TACTAAAAGTGACTCTCTAATTTCTATTTGGTAATT
AGTTTGCAGAGAAAGACAATATAGTTCTCT
TGATCACTCCACTGTTCATAGGGATCCATC
GTATCTATATTCATAGGAAACACCATT
ACATTTCTTCAGGGTGTCTGACTAA

25 *sub C2* 5. A set of allele specific primers for each of the following alleles of the CFTR gene: 621+1 G>T, R553X, G551D, R117H, R1162X and R334W *mutations*.

6. A set of primers as claimed in claim 5 and comprising the following diagnostic primer sequences:

GTATCTATATTCATCATAGGAAACACCACA
TGCCATGGGGCTGTGCAAGGAAGTATTGA
AGCCTATGCCTAGATAAATCGCGATAGACT
CCTATGCACTAATCAAGGAATCATCCTGT
GCTAAAGAAATTCTTGCTCGTTGTT
GACTGACTGACTGACTGACTCTGACTGACTTATTCA
CCTTGCTAAAGAAATTCTTGCTGA
TATTTTATTTCAGATGCGATCTGTGAGTT

7. A set of primers comprising the following diagnostic primer and amplification primer sequences:

TCTTGGGATTCAATAACTTGCACAGTCA
GAATTCCCAAACCTTTAGAGACATC
TACTAAAAGTGAECTCTAATTTCTATTTGGTAATT
AGTTGAGAGAAAGACAATATAGTTCT
TAATCTCTACCAAATCTGGATACTATACC
TGATCACTCCACTGTTCATAGGGATCCATC
AATTGAGAGAACTTGATGGTAAGTACA
GTATCTATATTCATCATAGGAAACACCATT
CCAGACTTCACCTCTAA~~TGATGATTATGGG~~
ACATTTCCCTTCAGGGTGTCTGACTAA
TTGGATCAAATTTCAGTTGACTTGTATC

8. A set of primers comprising the following diagnostic primer and amplification primer sequences:

GTATCTATATTCATCATAGGAAACACCACA
GACTTCACCTCTAATGATGATTATGGGAGA
TGCCATGGGGCTGTGCAAGGAAGTATTGA
AGCCTATGCCTAGATAAATCGCGATAGACT
GTTTCACATAGTGTATGACCCCTATATACACTCATT
CCTATGCACTAATCAAAGGAATCATCCTGT
TTTGGTTATTGCTCCAAGAGAGTCATACCA
GCTAAAGAAATTCTTGCTCGTTGTT
GACTGACTGACTGACTGACTCTGACTGACTTATTCA

CCTTGCTAAAGAAATTCTTGCTGA
TAAAATTGGAGCAATGTTGTTTGACC
TATTTTATTCAGATGCGATCTGTGAGTT
TTTGCTGTGAGATCTTGACAGTCATTT

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9. A set of primers as claimed in any one of the previous claims and comprising one or more of the following control primers:

GAGCACAGTACGAAAAACACCT
AAACTTTACAGGGATGGAGAACG
AGAGGATTATCTATGCAAATCCTTGTAAACC
TCAACTTCACTATCAAAGTCATCATCTAG

10. A diagnostic kit for detecting the presence or absence of twelve mutations in the cystic fibrosis transmembrane conductor regulator (CFTR) gene which comprises sets of primers as claimed in any of the previous claims.

one of claim 1-8

Add A
add B
add C
add D